



TAP1 gene

transporter 1, ATP binding cassette subfamily B member

Normal Function

The *TAP1* gene provides instructions for making a protein that plays an important role in the immune system. The TAP1 protein assembles with another protein called TAP2 (produced from the *TAP2* gene) to form a protein complex called transporter associated with antigen processing (TAP) complex. This complex, which is found in the membrane of a cell structure called the endoplasmic reticulum, moves (transports) protein fragments (peptides) from foreign invaders into the endoplasmic reticulum. There, the peptides are attached to major histocompatibility complex (MHC) class I proteins. The peptide-bound MHC class I proteins are then moved to the surface of the cell so that specialized immune system cells can interact with them. When these immune system cells recognize the peptides as harmful, they launch an immune response to get rid of the foreign invaders.

Health Conditions Related to Genetic Changes

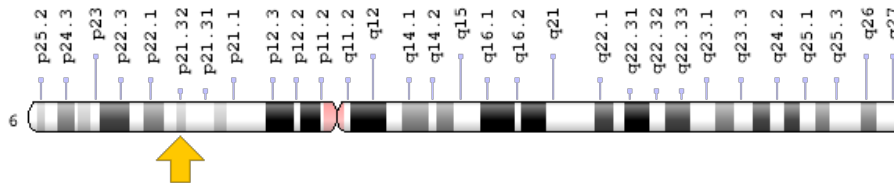
Bare lymphocyte syndrome type I

At least six mutations in the *TAP1* gene have been found to cause bare lymphocyte syndrome type I (BLS I). This immune system disorder often causes recurrent bacterial infections in the respiratory tract and open sores (ulcers) on the skin, although some people with BLS I have no symptoms of the condition. *TAP1* gene mutations involved in BLS I prevent production of functional TAP1 protein. Absence of functional TAP1 impairs the formation of the TAP complex, without which peptides from foreign invaders cannot be transported into the endoplasmic reticulum and attached to MHC class I proteins. Consequently, MHC class I proteins are broken down, which results in a shortage of these proteins on the surface of cells. A lack of MHC class I proteins impairs the body's immune response to bacteria, leading to recurrent bacterial infections. Researchers are unsure why people with BLS I do not also get viral infections, but they suspect that other immune processes are able to recognize and fight viruses. It is also not clear how *TAP1* gene mutations are involved in the development of skin ulcers.

Chromosomal Location

Cytogenetic Location: 6p21.32, which is the short (p) arm of chromosome 6 at position 21.32

Molecular Location: base pairs 32,845,209 to 32,853,704 on chromosome 6 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ABC transporter, MHC 1
- ABC17
- ABCB2
- APT1
- ATP-binding cassette sub-family B member 2
- ATP-binding cassette, sub-family B (MDR/TAP), member 2
- D6S114E
- peptide supply factor 1
- peptide transporter involved in antigen processing 1
- peptide transporter PSF1
- peptide transporter TAP1
- PSF-1
- PSF1
- RING4
- TAP1N
- transporter 1 ATP-binding cassette sub-family B
- transporter 1, ATP-binding cassette, sub-family B (MDR/TAP)

- transporter associated with antigen processing
- transporter, ATP-binding cassette, major histocompatibility complex, 1

Additional Information & Resources

Educational Resources

- Immunobiology: The Immune System in Health and Disease (fifth edition, 2001): The Major Histocompatibility Complex and Its Functions
<https://www.ncbi.nlm.nih.gov/books/NBK27156/>
- Molecular Biology of the Cell (fourth edition, 2002): T Cells and MHC Proteins
<https://www.ncbi.nlm.nih.gov/books/NBK26926/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TAP1%5BTIAB%5D%29+OR+%28transporter+1,+ATP+binding+cassette+subfamily+B+member%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- TRANSPORTER, ATP-BINDING CASSETTE, MAJOR HISTOCOMPATIBILITY COMPLEX, 1
<http://omim.org/entry/170260>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TAP1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TAP1%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:43
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:6890>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6890>
- UniProt
<https://www.uniprot.org/uniprot/Q03518>

Sources for This Summary

- Eggensperger S, Tampé R. The transporter associated with antigen processing: a key player in adaptive immunity. *Biol Chem*. 2015 Sep;396(9-10):1059-72. doi: 10.1515/hsz-2014-0320. Review. *Citation on PubMed*: <https://www.ncbi.nlm.nih.gov/pubmed/25781678>
- Hanalioglu D, Ayvaz DC, Ozgur TT, van der Burg M, Sanal O, Tezcan I. A novel mutation in TAP1 gene leading to MHC class I deficiency: Report of two cases and review of the literature. *Clin Immunol*. 2017 May;178:74-78. doi: 10.1016/j.clim.2017.01.011. Epub 2017 Feb 2. *Citation on PubMed*: <https://www.ncbi.nlm.nih.gov/pubmed/28161407>
- Neefjes JJ, Momburg F, Hämmerling GJ. Selective and ATP-dependent translocation of peptides by the MHC-encoded transporter. *Science*. 1993 Aug 6;261(5122):769-71. Erratum in: *Science* 1994 Apr 1;264(5155):16. *Citation on PubMed*: <https://www.ncbi.nlm.nih.gov/pubmed/8342042>
- Parcej D, Tampé R. ABC proteins in antigen translocation and viral inhibition. *Nat Chem Biol*. 2010 Aug;6(8):572-80. doi: 10.1038/nchembio.410. Review. Erratum in: *Nat Chem Biol*. 2010 Oct;6(10):782. *Citation on PubMed*: <https://www.ncbi.nlm.nih.gov/pubmed/20644544>
- OMIM: TRANSPORTER, ATP-BINDING CASSETTE, MAJOR HISTOCOMPATIBILITY COMPLEX, 1
<http://omim.org/entry/170260>
- Zimmer J, Andrès E, Donato L, Hanau D, Hentges F, de la Salle H. Clinical and immunological aspects of HLA class I deficiency. *QJM*. 2005 Oct;98(10):719-27. Epub 2005 Aug 8. Review. *Citation on PubMed*: <https://www.ncbi.nlm.nih.gov/pubmed/16087697>

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<https://ghr.nlm.nih.gov/gene/TAP1>

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